=> d his

(FILE 'HOME' ENTERED AT 16:22:40 ON 18 JUL 2006)

FILE 'MEDLINE, SCISEARCH, CAPLUS, BIOSIS' ENTERED AT 16:23:39 ON 18 JUL 2006

L1 140 S EFEMP1
L2 64 S FIBULIN-3
L3 10 S FBLN3
L4 181 S L1 OR L2 OR L3
L5 114 DUP REM L4 (67 DUPLICATES REMOVED)
L6 4 S L5 AND PY<=1999

=> d ti so au ab l6 2

L6 ANSWER 2 OF 4 MEDLINE on STN
TI A single **EFEMP1** mutation associated with both Malattia Leventinese and Doyne honeycomb retinal dystrophy.
SO Nature genetics, **(1999 Jun)** Vol. 22, No. 2, pp. 199-202. Journal code: 9216904. ISSN: 1061-4036.
AU Stone E M; Lotery A J; Munier F L; Heon E; Piguet B; Guymer R H; Vandenburgh K; Cousin P; Nishimura D; Swiderski R E; Silvestri G; Mackey D A; Hageman G S; Bird A C; Sheffield V C; Schorderet D F
AB Malattia Leventinese (ML) and Doyne honeycomb retinal dystrophy (DHRD) refer to two autosomal dominant diseases characterized by yellow-white deposits known as drusen that accumulate beneath the retinal pigment epithelium (RPE). Both loci were mapped to chromosome 2p16-21 (refs 5,6) and this genetic interval has been subsequently narrowed. The importance of these diseases is due in large part to their close phenotypic similarity to age-related macular degeneration (AMD), a disorder with a strong genetic component that accounts for approximately 50% of registered blindness in the Western world. Just as in ML and DHRD, the early hallmark of AMD is the presence of drusen. Here we use a combination of positional and candidate gene methods to identify a single non-conservative mutation (Arg345Trp) in the gene **EFEMP1** (for EGF-containing fibrillin-like extracellular matrix protein 1) in all families studied. This change was not present in 477 control individuals or in 494 patients with age-related macular degeneration. Identification of this mutation may aid in the development of an animal model for drusen, as well as in the identification of other genes involved in human macular degeneration.